



MLC1 gene

megalencephalic leukoencephalopathy with subcortical cysts 1

Normal Function

The *MLC1* gene provides instructions for making a protein that is found primarily in the brain but also in the spleen and white blood cells (leukocytes). Within the brain, the MLC1 protein is found in astroglial cells, which are a specialized form of brain cells called glial cells. Glial cells protect and maintain other nerve cells (neurons). The MLC1 protein functions at junctions that connect neighboring astroglial cells. The role of the MLC1 protein at the cell junction is unknown, but research suggests that it may control the flow of fluids into cells or the strength of cells' attachment to one another (cell adhesion). Studies indicate that the MLC1 protein may be involved in transporting molecules across the blood-brain barrier and the brain-cerebrospinal fluid barrier. These barriers protect the brain's delicate nerve tissue by allowing only certain substances to pass into the brain.

Health Conditions Related to Genetic Changes

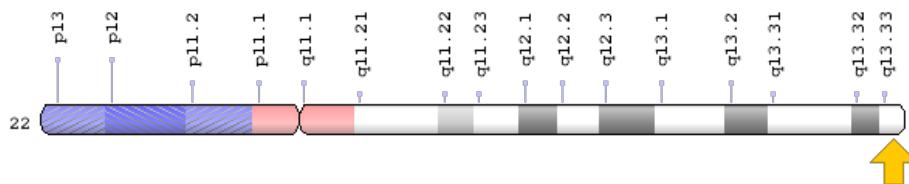
megalencephalic leukoencephalopathy with subcortical cysts

More than 80 mutations in the *MLC1* gene have been found to cause megalencephalic leukoencephalopathy with subcortical cysts type 1; this type accounts for 75 percent of all cases. This condition affects brain development and function, resulting in problems with movement and recurrent seizures. Most of the *MLC1* gene mutations that cause this condition change single protein building blocks (amino acids) in the MLC1 protein. These changes alter the structure of the MLC1 protein or prevent the cell from producing any protein. It is unknown how a lack of MLC1 protein at astroglial cell junctions impairs brain development and function, causing the signs and symptoms of megalencephalic leukoencephalopathy with subcortical cysts type 1.

Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 50,059,391 to 50,085,929 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA0027
- LVM
- megalencephalic leukoencephalopathy with subcortical cysts 1 gene product
- MLC
- MLC1_HUMAN
- VL

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): The Blood-Brain Barrier Undergoes Development
<https://www.ncbi.nlm.nih.gov/books/NBK28180/#A2335>
- Basic Neurochemistry (sixth edition, 1999): Virtually Nothing Can Enter or Leave the Central Nervous System Parenchyma Without Passing Through an Astrocytic Interphase
<https://www.ncbi.nlm.nih.gov/books/NBK28217/#A32>
- Molecular Biology of the Cell (fourth edition, 2002): Cell Junctions
<https://www.ncbi.nlm.nih.gov/books/NBK26857/>

GeneReviews

- Megalencephalic Leukoencephalopathy with Subcortical Cysts
<https://www.ncbi.nlm.nih.gov/books/NBK1535>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MLC1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MLC1 GENE
<http://omim.org/entry/605908>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MLC1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17082
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23209>
- UniProt
<http://www.uniprot.org/uniprot/Q15049>

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